

Table 2: organ system involvement in POLG-1 related mitochondrial disorders

Organ system	Symptom
General	Failure to thrive
Central Nervous System	Hypotonia Migraine Stroke like episodes Seizures especially myoclonic and epilepsy partialis continua Ataxia Chorea Delayed ocular movements Encephalopathy Parkinsonism Depression Psychosis Dementia Visual hallucinations
Peripheral nervous system	
Musculoskeletal	Ptosis External ophthalmoparesis Proximal myopathy Exercise intolerance Peripheral neuropathy
Eye	Cataract Retinopathy Cortical blindness
Ear	sensorineural hearing loss

Endocrine

Diabetes  
Premature ovarian failure  
Primary testicular failure

Gastrointestinal

Liver Dysfunction or failure of  
childhood or adult onset  
Dysmotility

Cardiac

Cardiomyopathy